

PARENT FACT SHEET

DISORDER

Isobutyryl-CoA Dehydrogenase Deficiency (IBD)

CAUSE

Isobutyryl-CoA Dehydrogenase (IBD) deficiency is caused when an enzyme called “isobutyryl-CoA dehydrogenase” is missing or not working properly. This enzyme is needed to break down proteins from the food we eat into other substances that are need for growth, development and energy.

IF NOT TREATED

IBD deficiency is very rare and little is known about the effects. Most people with IBD deficiency do not have signs or symptoms. A few children have developed a weakened and enlarged heart, weak muscle tone, and some developmental delays.

TREATMENT OPTIONS

Your baby’s primary doctor will work with a metabolic specialist and dietician to provide care for your child. The following treatments may be recommended for some babies:

- Medications: Some children benefit from taking a supplement called L-carnitine.
- Avoid letting the baby go long periods without eating. Your metabolic doctor and dietician will tell you how often your child needs to eat
- A special diet may be ordered for your child that is low in protein. The dietician will tell you if you should supplement your child’s diet with specially made medical foods.

IF TREATED

There is very little information available; however it is thought that with careful treatment, children with IBD deficiency will have normal growth and development. Treatment with Carnitine may improve heart function, anemia and may improve growth.